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Christ – Siemens – Touraine syndrome with a rare finding of Macroglossia - A case report of three siblings

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ABSTRACT:
Christ-Siemens-Touraine syndrome is one among about 150 types of Ectodermal dysplasia. It is the hypohidrotic type of Ectodermal dysplasia, usually inherited as X-Linked recessive type. It is considered to be a triad of hypodontia, hypotrichosis and hypohidrosis. Here we present three classical cases of this rare syndrome who were all siblings. Management of this disorder with special insight on dental considerations is highlighted.

Key words: Ectodermal dysplasia, Macroglossia, Hypodontia, Hypotrichosis, Hypohidrosis, Over dentures

INTRODUCTION

Ectodermal dysplasia syndrome is a large, heterogenous group of inherited disorders usually resulting from defective development of structures of ectodermal origin. The tissues primarily affected are the skin, hair, nails, eccrine glands and teeth. Hypohidrotic ectodermal dysplasia (HED), also known as Christ – Siemens – Touraine syndrome, is the most common phenotype in this group [1,2]. and is usually inherited as an X-linked recessive trait[3]. It is considered to be a triad of hypodontia, hypotrichosis and hypohidrosis [3].

CASE REPORT

Three patients who were siblings (Fig 1), reported to our department with the chief complaint of multiple missing teeth since childhood. On clinical examination, we noted the following findings in all the three patients: The skull was found to be inverted triangle in shape (fig 2). Marked frontal bossing, depressed nasal bridge and obliquely inserted ears, causing them to stand out (Fig. 3), were also noted. Hypohidrosis with intolerance to heat was present in all the three patients. They had a peculiar habit of pouring water all over the body to counter the heat. Hypohidrosis was demonstrated using Minor's starch iodine test (Fig.4).

To demonstrate the presence of hyperpyrexia, we recorded the body temperature of all the three patients using clinical thermometer. All of them had a body temperature of 99˚ F which was above normal body temperature (Table 1).

The scalp hair was found to be sparse, blond, fine, and short (Fig. 2). Hair on the eyebrows and eyelashes were also found to be sparse (Fig.5). The body was devoid of lanugo hair. The finger and toe nails were normal.

Extraorally, lips were protuberant. The vermilion border was indistinct and pseudorhagades were present (Fig.6). The most striking intraoral manifestation was hypodontia. Those teeth that were present were conical in shape and microdontic. Case 1 is under prosthodontic treatment. The alveolar ridge was poorly developed due to the absence of teeth (Fig. 7). We also noted a peculiar finding that was not reported in earlier literature; Macroglossia with fissured tongue was present in all the three siblings (Fig.8).

Radiographic examination included Intraoral periapical radiograph, Panoramic radiograph and lateral cephalogram. Intraoral periapical radiograph revealed cone shaped maxillary incisors with small pulp chambers (Fig.9). Panoramic radiograph revealed hypodontia, cone shaped maxillary anterior teeth and resorbed mandibular ridge (Fig.10). Lateral cephalogram revealed frontal bossing, depressed nasal bridge and resorbed mandibular ridge (Fig 11).

With the help of family history we drew the pedigree chart (Fig.12). Both the parents were apparently healthy and not affected by this disease. But all the three male offsprings were affected by the disease, denoting that the offsprings would have got the recessive X-chromosome from their mother who is a carrier. This implies that it is an X-linked recessive disorder.

On correlating with history and clinical & radiographic findings, we arrived at the final diagnosis of Christ-Siemens-Touraine syndrome.

Dental management of these patients includes managing xerostomia and hypodontia. Artificial saliva was prescribed to overcome xerostomia. Chlorhexidine mouthwash 0.2% was prescribed to handle increased incidence of infection due to xerostomia.

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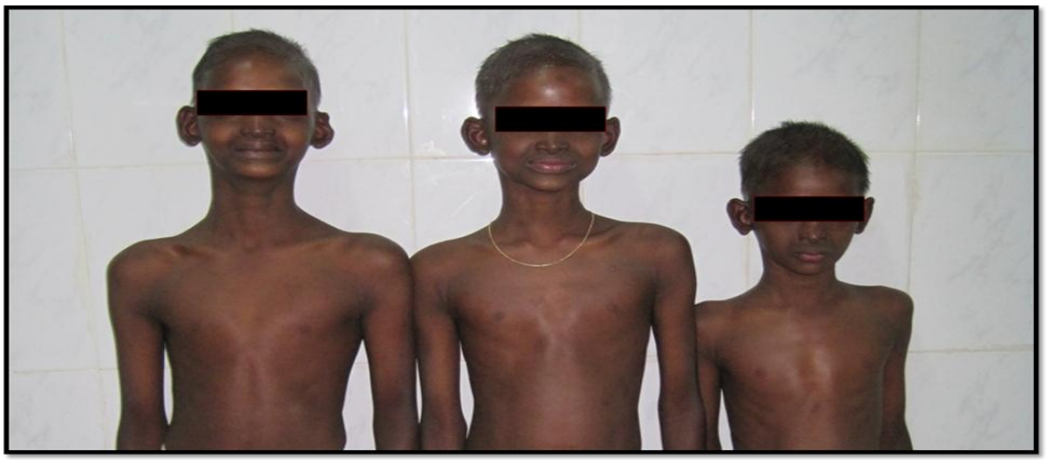
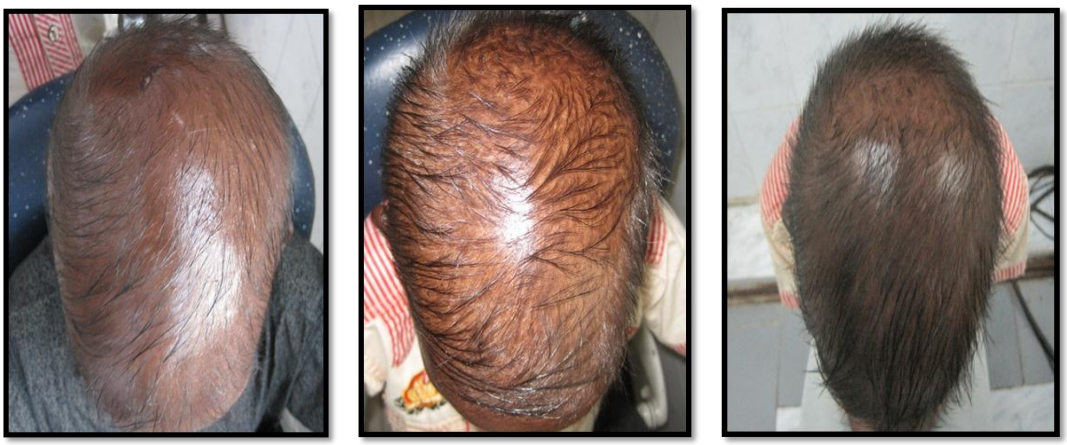
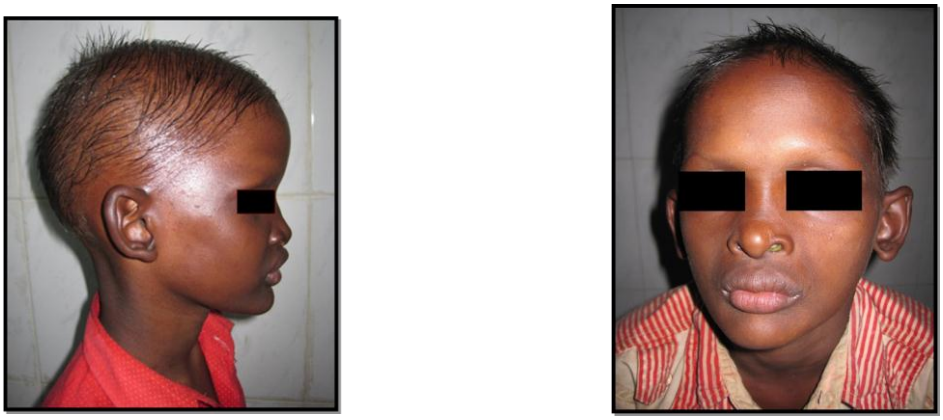


Fig. 1 The three siblings



CASE 1 CASE 2 CASE 3

Fig. 2



PROFILE VIEW FRONTAL VIEW

Fig. 3

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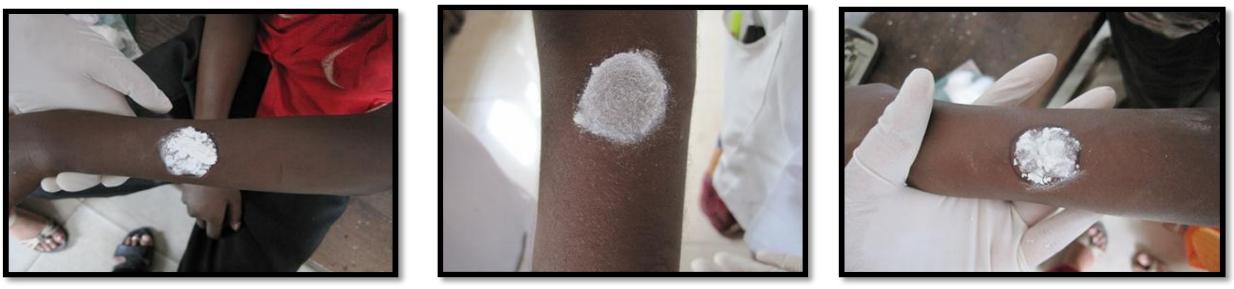
PATIENT	BODY TEMPERATURE (Degree F)
Case 1	99
Case 2	99
Case 3	99

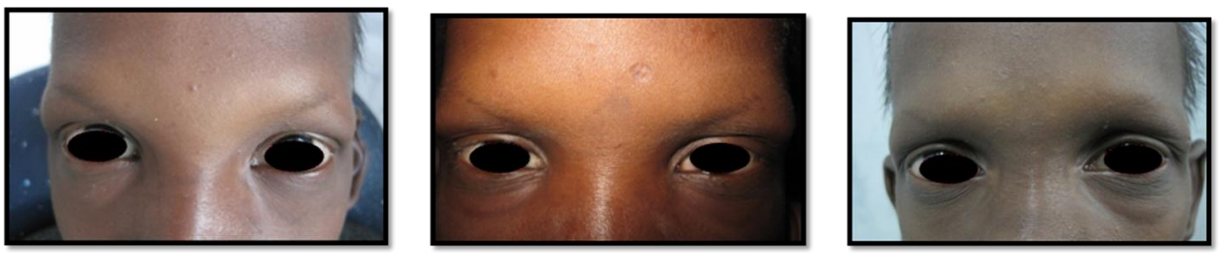
Table 1 Body Temperature recordings demonstrating the presence of hyperpyrexia



CASE 1 CASE 2 CASE 3

CONTROL

Fig. 4 – Minor's Starch Iodine test: Markings were made on the forearm of all the three patients and a normal control. Then Iodine solution was painted over the area with a paint brush. After a period of one hour, Corn starch powder was sprinkled over that area. There was no color change in all the three patients, but a purple color change was noted on the control [11, 12].



CASE 1 CASE 2 CASE 3

Fig. 5 Sparse eyebrows and eye lashes with hypertelorism

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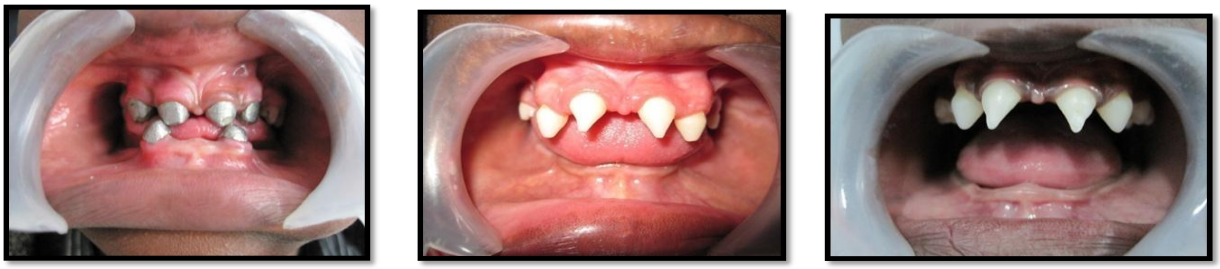
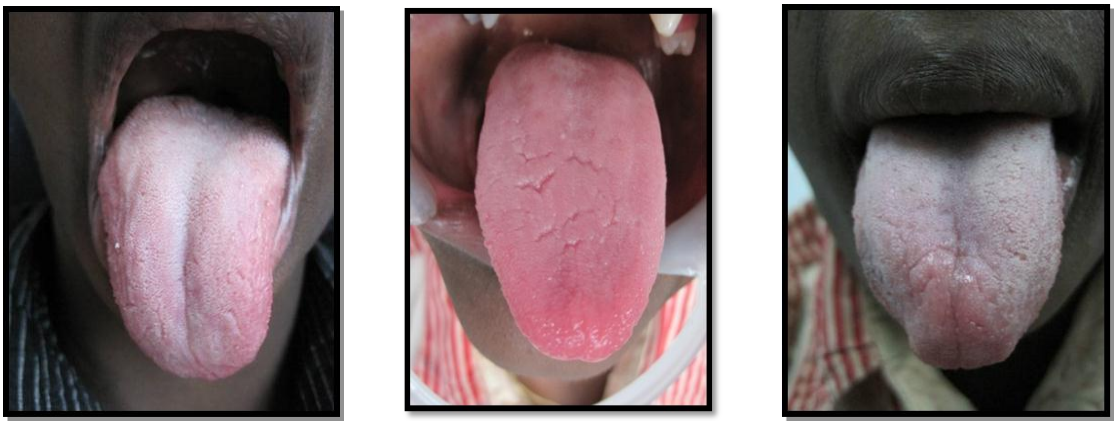
CASE 1 CASE 2 CASE 3

Fig. 6 Protuberant lips with pseudorhagades



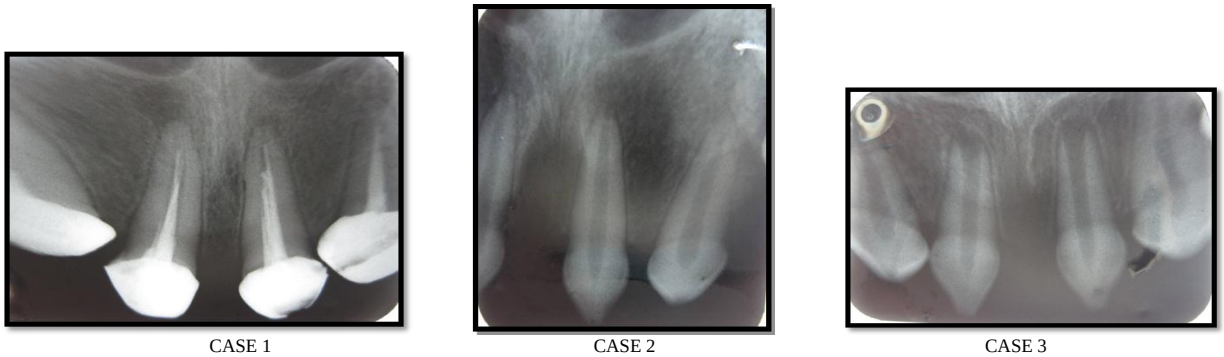
CASE 1 CASE 2 CASE 3

Fig. 7 Hypodontia, cone shaped anterior teeth and resorbed alveolar ridge



CASE 1 CASE 2 CASE 3

Fig. 8 Macroglossia with fissured tongue



CASE 1 CASE 2 CASE 3

Fig. 9 Intraoral periapical radiograph (IOPA): Reveals cone shaped maxillary incisors with small pulp chambers

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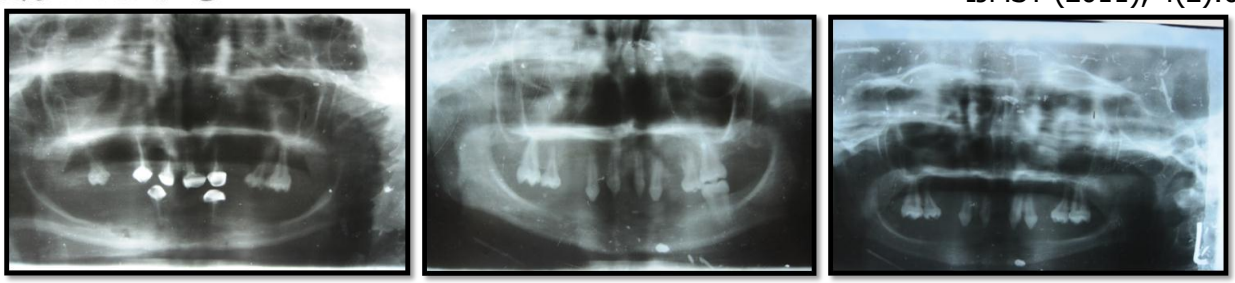
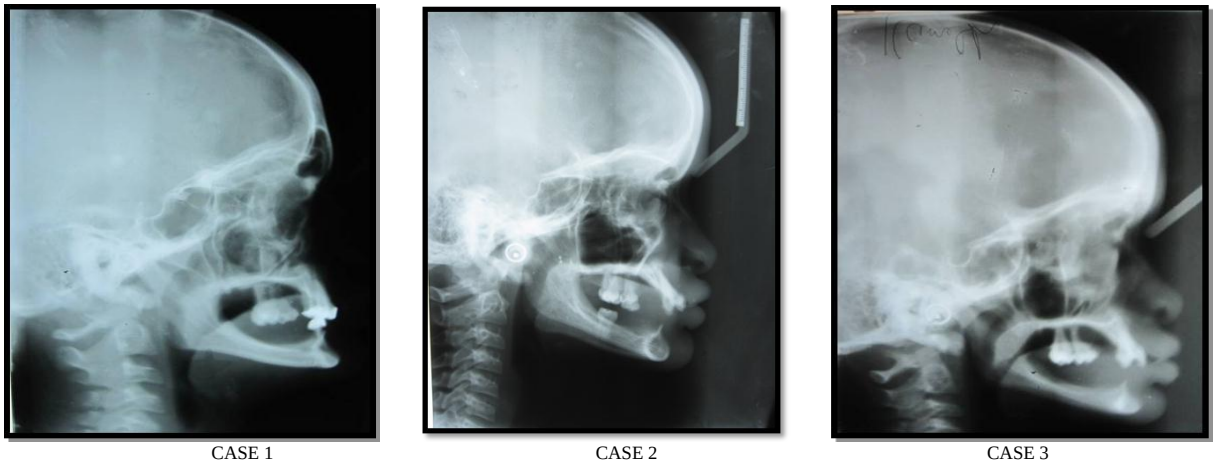


CASE 1 CASE 2 CASE 3

Fig. 10 Panoramic radiograph (OPG): Reveals hypodontia, cone shaped maxillary anterior teeth and resorbed mandibular ridge



CASE 1 CASE 2 CASE 3

Fig. 11 Lateral cephalogram: revealed frontal bossing, depressed nasal bridge and resorbed mandibular ridge

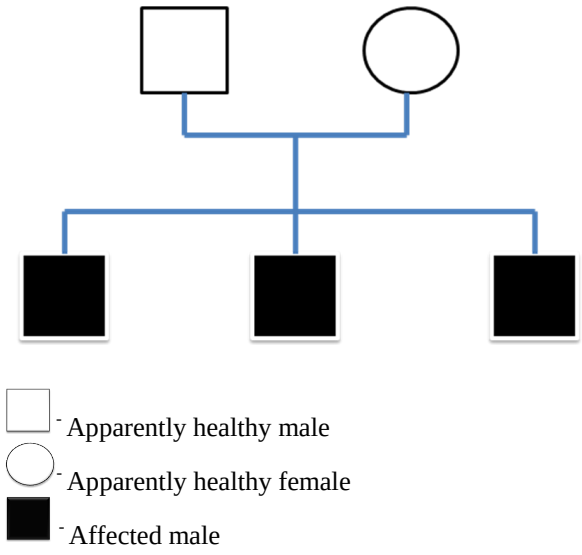


Fig. 12 Pedigree Chart

With respect to prosthetic rehabilitation, it was planned to give over denture wherever feasible and complete denture in other situations. Over denture will improve the retention which seems to be compromised in these patients. It prevents further resorption of the residual alveolar ridge and maintains proprioceptive responses. For case 1, intensional Root canal treatment was done for maxillary and mandibular anterior teeth and metal copings were placed (Fig.7, 9). Then over dentures were fabricated for both the arches. For case 2, over dentures were fabricated for both the arches without coping for the abutments as they had sufficient crown length. For case 3, over denture was given for maxillary arch and complete denture for mandibular arch. The patients are under close follow-up. Implant-supported prosthesis was not opted for these patients due to their young age, continuing growth of the jaws and insufficient alveolar bone support.

Later, the patients were referred to the dermatologist for further management.

DISCUSSION

Hereditary ectodermal dysplasia is a rare group of inherited disorders characterized by aplasia or dysplasia of tissues of ectordermal origin, such as hair, nails, teeth and skin. It was first reported by Thurman in 1848 [5, 6]. The condition is thought to occur in approximately 1 in every 100,000 live births [2, 4, 5]. Freire – Maia and Pinheiro described 154 varieties of ectodermal dysplasia with multiple combinations of abnormal ectodermally derived structures [7], the most common being X-Linked HED (Christ-Siemens-

Touraine syndrome) [1, 2]. It is placed under Group A, subgroup 1-2-3-4 in Pinheiro and Freire-Maia classification [7].

The phenotype of X-Linked HED includes sparse scalp hair, largely absent body hair, deficiency of eccrine sweat glands, and anodontia or oligodontia with conical teeth. The facies is distinctive with prominence of the forehead, a depressed nasal bridge, prominent lips, and periorbital wrinkling and pigmentation [5, 8, 9]. Hypodontia was extensive, but there was relative sparing of upper first molar, upper central incisor and both canines [10]. These findings were present in all our three cases.

Deficiency of eccrine sweat glands leads to hypohidrosis and hyperpyrexia. We used Minor's Starch iodine test to demonstrate hypohidrosis [11, 12]. There was normal sweat secretion in the control, which aided the iodine solution to be in wet state without drying, in turn helping it to react with Corn Starch powder to produce the color change. But this was absent in all the three patients, confirming the presence of hypohidrosis.

In addition to these findings, we noted macroglossia to be present in all the three siblings. This is a peculiar finding which was not reported in earlier literatures. Macroglossia may be due to compensatory hyperplasia of the tongue musculature to hypodontia and resorbed alveolar ridge. Fissuring of the tongue was also present which may be due to decrease in the number of minor salivary glands in turn causing xerostomia [12].

Pedigree chart confirms that it is an X-Linked recessive disorder. According to the clinical – genetic classification of Maneula Priolo and Carmelo Lagana, X-Linked HED is placed under Group 1 which includes disorders in which a defect in developmental regulation and in epithelial – mesenchymal interaction can be recognized or hypothesized on the basis of an identified causative gene, its putative of proven function, and pattern of expression [6]. The EDA 1 gene responsible for X-Linked HED has been mapped to Xq12-q13 [2, 3], which codes for Ectodysplasin-A1 protein [6]. The defect leads to altered epithelial – mesenchymal interaction, an important mechanism in the pathogenesis of ectodermal dysplasia [6].

Management is directed at optimizing psychological development, establishing optimal oral function, and preventing hyperthermia [3]. Wigs can be used to manage hypotrichosis. To counteract hypohidrosis, patients can be advised to drink adequate amount of cool water and maintain cool environment by using air conditioning, spray bottle of water or wet clothing. All the family members must be given genetic counseling.

Dental considerations

- To correct hypodontia, removable and/or fixed partial denture, over denture, complete denture and implant-retained prosthesis are the various options available. The clinician can have his choice from any of these options in various combinations depending on the clinical situations, so as to optimize the function and aesthetics. Because of the early age at intervention and the need to easily modify the intraoral prosthesis during rapid growth periods, a removable partial denture or complete denture prosthesis is indicated initially. Implant-retained prosthesis can be used at a later age when the growth is complete. Hypodontia is associated with lack of development of the alveolar ridge and results in less volume of bone available to support a conventional prosthesis. As a result, the placement of endosseous implants in locations favourable for subsequent restorations may be difficult [13].
- The complete or partial dentures delivered need to be periodically replaced every 2.5 years to compensate for the resorption of alveolar ridges [3].
- Consideration must be given when implants are placed in partially edentulous ridge, as it might result in submergence of the implant due to the continued increase in alveolar height caused by eruption of adjacent natural teeth [4].
- To counteract xerostomia, artificial saliva can be used.

In conclusion, early intervention is important to manage hypodontia/anodontia to ensure physiological and psychological well being. This necessitates a multi-disciplinary approach involving the Oral medicine and Radiologist, pedodontist, prosthodontist, endodontist, dermatologist, paediatrician and psychologist. Moreover, successful management of these patients requires good co-operation and communication among the patient, his parents and the dental team. Though there could be many variations to this disorder and as we are continuing to diagnose more such patients as part of the study, we could say from this case report that we could successfully manage the Christ-Siemens-Touraine syndrome.

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